## BIOGRAPHICAL SKETCH

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NAME: Scholz, Sonja Waltraud

eRA COMMONS USER NAME (credential, e.g., agency login): sscholz5

POSITION TITLE: Tenure Track Investigator, National Institute of Neurological Diseases and Stroke; Adjunct Assistant Professor of Neurology, Johns Hopkins University

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable. Add/delete rows as necessary.)*

| INSTITUTION AND LOCATION | DEGREE *(if applicable)* | Completion Date MM/YYYY | FIELD OF STUDY |
|---|---|---|---|
| Medical University Innsbruck, Austria | M.D. | 10/2004 | Medicine/Neuroscience |
| National Institute on Aging, Bethesda, MD, USA | (Post-Doc.) | 11/2009 | Human Genetics |
| University College London, United Kingdom | Ph.D. | 04/2010 | Neurogenomics |
| Georgetown University, Washington, DC, USA | (Post-Doc.) | 06/2011 | Neuroscience |
| Johns Hopkins University, Baltimore, MD, USA | (Intern) | 06/2012 | Internal Medicine |
| Johns Hopkins University, Baltimore, MD, USA | (Resident) | 06/2015 | Neurology |
| Johns Hopkins University, Baltimore, MD, USA | (Fellow, adjunct) | 10/2018 | Neurodegeneration |

### A. Personal Statement

I am a board-certified neurologist and scientist with a career-long interest in the genetics of neuro-degeneration, particularly movement disorders. I am the chief of the Neurodegenerative Diseases Research Unit at the U.S. National Institutes of Health. In my research program, we specialize in applying modern genomic techniques and data-driven approaches to assess molecular genetic mechanisms implicated in complex neurological syndromes. We aim to use this knowledge to improve pathobiologic understanding and diagnostic accuracy. To date, my research has had significant impact on our understanding of several neurological diseases, including the discovery of genes underlying dystonia, Parkinson's disease, spinocerebellar ataxia, amyotrophic lateral sclerosis, frontotemporal dementia, and multiple system atrophy. My research has provided crucial insights into the pathogenesis of Lewy body dementia and its molecular relationships to Alzheimer's disease and Parkinson's disease. I plan to continue to use state-of-the-art genomic tools to enhance our understanding of these devastating conditions and to identify targets suitable for disease-modifying, therapeutic interventions.

### B. Positions and Honors

**Positions and Employment**

2005 - 2009 **Postdoctoral Fellow**, Laboratory of Neurogenetics, National Institute on Aging, Bethesda, MD
2009 - 2011 **Postdoctoral Fellow**, Department of Neuroscience, Georgetown University, Washington, DC
2011 - 2012 **Intern**, Osler Program, Department of Medicine, Johns Hopkins University, Baltimore, MD
2012 - 2015 **Resident**, Department of Neurology, Johns Hopkins University, Baltimore, MD
2015 - 2018 **Assistant Clinical Investigator**, National Institute of Neurological Disorders and Stroke, Bethesda, MD; adjunct fellow specializing in neurodegeneration at Johns Hopkins University
2018 - present **Lasker Clinical Research Scholar Tenure Track Investigator**, National Institute of Neurological Disorders and Stroke, Bethesda, MD
2018 - present **Adjunct Assistant Professor of Neurology**, Johns Hopkins University, Baltimore, MD

**Other Experience and Professional Memberships**

- Professional Memberships
  Member, American Academy of Neurology
  Member, American Neurological Association
  Member, Movement Disorders Society
  Member, Movement Disorders Society Multiple System Atrophy Study Group (MODIMSA)
  Member, Movement Disorders Society Progressive Supranuclear Palsy Study Group
  Member, Alpha Omega Alpha Honor Medical Society
  Member, American Society of Human Genetics
- *Ad hoc* peer reviewer
  Neurology, Brain, JAMA Neurology, Annals of Neurology, Movement Disorders, Neurobiology of Aging, Lancet Neurology and several other journals
- F1000Prime: Faculty Member for Neurological Disorders Section
- Editorial board member: Journal of Parkinson's Disease
- Teaching: I give lectures at the following courses:
  Neuro-degenerative Disorders (Course # 330.802.01), Johns Hopkins University
  Diseases & Disorders of the Nervous System (Course # AS.080.360), Johns Hopkins University
- Medical Licenses, Certifications
  Licensed Physician, Medical Board of Maryland (D0078869)
  Diplomate of the American Board of Psychiatry and Neurology

**Honors**

| | |
|---|---|
| 2004 | Poster prize, Austrian Parkinson Society meeting, Pörtschach, Austria |
| 2007 / 2008 | Development in Molecular Science, studentship by the University College London (UK) |
| 2008 / 2009 | Development in Molecular Science, studentship by the University College London (UK) |
| 2009 | Travel Fellowship Award for the 134th Annual Meeting of the American Neurological Association, Baltimore, MD, USA |
| 2015 | McFarland Transition to Independence Award, NINDS, National Institutes of Health |
| 2018 | Lasker Clinical Research Scholar Award, NINDS, National Institutes of Health |

**C. Contributions to Science**

1. *In vivo* monitoring of neurological disease and therapeutic interventions. Gene therapy holds great promise for the treatment of diseases involving the central nervous system. For this reason, my early research as part of my medical doctorate studies focused on functional imaging of a Huntington's disease rat model that would allow us to monitor therapeutic interventions, such as stem cell grafting or gene therapy. During this research, I developed a sensitive model system for *in vivo* monitoring using pinhole SPECT and MRI co-registration. This methodology is considerably cheaper than small-animal PET-imaging and it is superior to commonly used behavioral testing.
   a) Scherfler C, **Scholz SW**, Donnemiller E, Decristoforo C, Oberladstätter M, et al. (2005). Evaluation of [123I]-IBZM pinhole SPECT for the detection of striatal dopamine D2 receptor availability in rats. *NeuroImage*, 24(3): 822-31.

2. Discovering genetic variation in human health and neurological disease. During my early course of research, it became apparent to me that the genetic etiology of neurodegenerative diseases has to be further elucidated to provide us with robust targets for therapeutic interventions. My PhD in neurogenomics therefore focused on applying modern genomic technologies to dissect risk genes implicated in a variety of neurological diseases and to explore normal variation in diverse human populations. My research led to the discovery of several new disease genes, highlighting the critical role that genetic variability plays in the pathogenesis of neurological disease.
   a) Jakobsson M, **Scholz SW (joint first),** Scheet P, Gibbs JR, et al. (2008). Genotype, haplotype and copy-number variation in worldwide human populations. *Nature*, 451(7181): 998-1003.

b) Camargos S, **Scholz S (joint first),** Simon-Sanchez J, Paisan-Ruiz C, et al. (2008). DYT16, a novel young-onset dystonia-parkinsonism disorders: identification of a segregating mutation in the stress-response protein PRKRA. *Lancet Neurology*, 7(3):207-15.
c) **Scholz SW,** Houlden H, Schulte C, Sharma M, et al. (2009). SNCA variants are associated with increased risk for multiple system atrophy. *Annals of Neurology*, 65(5): 610-4.

3. Investigation of genetic risk driving neurodegenerative parkinsonism syndrome. Atypical parkinsonism syndromes are complex, underserved neurological diseases of unknown etiology. Our understanding of genetic risk factors in the pathogenesis of these devastating disorders indicate intricate molecular relationships between neurodegenerative entities. With this in mind, I established a laboratory at the National Institutes of Health with the primary mission being the exploration of genetic risk factors in atypical parkinsonism syndromes and their molecular relationships. For example, we have already shown that Lewy body dementia has a strong genetic predisposition and that the genetic risk profile of this unusual dementia syndrome falls along a spectrum between the profiles seen Parkinson's disease and Alzheimer's disease. The ultimate goal of my research is to incorporate molecular knowledge into our diagnostic, prognostic and treatment approaches towards managing these debilitating neurological diseases.
    a) Blauwendraat C, Pletnikova O, Geiger JT, Murphy NA, ... **Scholz SW** (2019). Genetic analysis of neurodegenerative diseases in a pathology cohort. *Neurobiology of Aging*, 76:214.
    b) Sailer A, **Scholz SW**, Nalls MA, Schulte C, Federoff M, ... Houlden H. (2016). A genome-wide association study in multiple system atrophy. *Neurology*, 87(15):1591-1598.
    c) Geiger JT, Ding J, Crain B, Pletnikova O, … **Scholz SW** (2016). Next-generation sequencing reveals substantial genetic contribution to dementia with Lewy bodies. *Neurobiology of Disease*, 94:55-62.

**Complete List of Published Work in MyBibliography (h-index = 29):**
https://www.ncbi.nlm.nih.gov/myncbi/browse/collection/51642443/?sort=date&direction=descending

**D. Additional Information: Research Support and/or Scholastic Performance**

**Ongoing Research Support**

Intramural grant by the National Institute of Aging — Scholz (Co-PI) 03/01/2018 - present
- Project title: Genome sequencing of Lewy body dementia and frontotemporal dementia: a public resource for the research community.
- Description: The objectives of this project are to (1) extend modern gene discovery methods to Lewy body dementia and frontotemporal dementia and (2) to generate a unique resource for the scientific community that will accelerate the pace of discovery in these underserved diseases.
- Role: Co-PI

Lasker Clinical Research Scholar Award — Scholz (PI) 09/01/2018 - present
- Granting organization: National Institutes of Health/NINDS
- Project title: Genomic characterization of atypical parkinsonism: a precision medicine roadmap.
- Description: The aim of this research program is to advance molecular genetic understanding of Lewy body dementia and related parkinsonism syndromes.
- Role: PI

**Completed Research Support**

Rapid Response Innovation Award — Scholz (PI) 2010 - 2011
- Granting organization: Michael J. Fox Foundation
- Project title: SNCA haplotyping in Parkinson disease and multiple system atrophy
- Description: The goal of this project was to perform targeted next-generation sequencing of the SNCA locus in pathologically confirmed cases with multiple system atrophy, Parkinson disease and normal controls to study the haplotype structure at this critical region.
- Role: PI

R25 Research Education Grant — Hillis-Trupe (PI) — 2014 - 2015

- Granting organization: National Institutes of Health/NINDS (NS065729)
- Project title: Exome sequencing in dementia with Lewy bodies
- Description: This is a mentored research education program that granted protected research time during residency training to facilitate a transition to an academic, neurologist-neuroscientist career pathway.
- Role: Trainee

McFarland Transition to Independence Award — Scholz (PI) — 08/09/2015 - 08/30/2018

- Granting organization: National Institutes of Health/NINDS (NS03154)
- Project title: The genetics of atypical parkinsonism
- Description: The goal of this mentored, intramural career development grant was to apply modern genomic technologies to study the genetic risk genes implicated in atypical parkinsonism syndromes and to facilitate a transition to an independent physician-scientist career pathway.
- Role: PI